
Lab Dept: Anatomic Pathology

Test Name: **OSTEOGENESIS IMPERFECTA
(COL1A1/COL1A2) SEQUENCING**

General Information

Lab Order Codes: COL10

Synonyms: Brittle Bone Disease; Osteogenesis Imperfecta Types I, IIA, III, IV

CPT Codes: 81408 x2 – Molecular Pathology procedure, Level 9

Test Includes: Using genomic DNA, the coding regions of the COL1A1/COL1A2 genes are amplified using flanking intron primers and sequenced by automated sequencing.

Logistics

Test Indications: For confirmation of the clinical diagnosis of osteogenesis imperfecta (OI), when identification of a gene mutation is needed for prenatal diagnosis of other family members or in the investigation or in the investigation of OI as a possible diagnosis in with “unexplained fractures”.

Collagen I is a tough, fibrous protein that provides a major part of the strength of bones. Mutations in two genes that code for collagen (COL2A1, COL1A2) cause either decreased synthesis of the protein or cause synthesis of defective forms of the protein. The result is weak or fragile bones. Mutations in the two genes have been found in over 90% of the patients with osteogenesis imperfecta (OI) and have been shown to be the cause of disease in these patients.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: CTGT

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 6 - 8 weeks

Special Instructions: [See Patient Preparation](#). Request special forms for patient consent, physician signature and clinical history from Children’s Laboratory or print [Request form](#) here.

Note: These completed forms must accompany the specimen or come with the patient when they present at the lab to be drawn.

Specimen

Specimen Type: Whole blood

Container: Lavender top (EDTA) tube

Draw Volume: 6 mL (Minimum: 4 mL for infants) blood

Processed Volume: Same as Draw Volume

Collection: Routine venipuncture

Special Processing: Lab Staff: **Do Not** process. Specimen should remain as whole blood in original collection container. Send Sunday – Thursday (to arrive Monday-Friday) via overnight shipping at room temperature. Include completed requisition and patient history with the specimen. Forward promptly.

Note: For specimens collected Friday – Saturday (or on a holiday), they should be held in Children’s Laboratories at refrigerated temperatures and shipped on Sunday if possible (or the next business day Monday – Thursday).

Patient Preparation: None

Sample Rejection: Mislabeled or unlabeled specimen; frozen specimen

Interpretive

Reference Range: No mutations detected

Critical Values: N/A

Limitations: The sensitivity for identifying a gene mutation by genomic sequencing of COL1A1/COL1A2 in an individual with OI is approximately 95%. Sequencing of type I collagen genes is slightly more sensitive than collagen screening in this regard.

Methodology: DNA sequencing

References: [Connective Tissue Gene Tests](#) March 2012
(484) 224-2900 Fax (484) 244-2904

Updates:

4/13/2010: Test moved from Tulane University to University of Washington.

8/13/2012: Moved from University of Washington to CTGT.

2/7/2013: CPT update

7/10/2013: CPT update CTGT, previously listed as 81408x1.